

Live birth prevalence of spina bifida in a tertiary hospital of south India- A 6-year survey.

Dr P. Sankar

Associate professor

Department of Neurosurgery

Dharmapuri Medical College and Hospital

Chennai, India

Dr Virender Gombra

Professor

Department of Oral Medicine and Radiology

Faculty of Dentistry

Jamia Millia Islamia

New Delhi, India

Dr Kaveri C

Assistant professor

Department of Neurosurgery

Dharmapuri Medical College and Hospital

Chennai, India

Corresponding author: Dr R Thillai Karasi

Assistant Professor

Department of Dental surgery

Dharmapuri Medical College and Hospital

Chennai, India

Abstract:

Objective: The study was undertaken to estimate the prevalence of the spina bifida at a South Indian tertiary hospital in six-year duration. **Material and methods:** The study was conducted in Government Dharmapuri Medical College Hospital (GDMCH), Tamil Nadu, South India. Data were obtained retrospectively by reviewing the patients' records from the department of paediatrics GDMCH. The overall number of the live births was recorded from 2015-2020. The patients' parents were contacted telephonically for collecting the patient's details such as the history of consanguineous marriage, age of the parents at the time of pregnancy and health status to check the deceased patient's number in six years duration. **Results:** Live birth number from 2015-2020 at the hospital was 51364. There were 66 babies born with Spina bifida. The live birth prevalence of spina bifida for the present study is 1.28/1000 births in six years duration with almost equal number of male and female ratio.

Conclusion: Spina bifida is one of the common congenital neural tube defects, which may cause stillbirth or severe morbidities in the affected individual. Regular assessment of the incidence will help in evaluating the requirement of further research for the same. Preventive measures like mandatory preconceptional folic acid fortification during pregnancy are required to decrease the prevalence of spina bifida and their comorbidities.

Keywords: Neural tube defects (NTDs), meningomyelocele, Myelomeningocele, congenital abnormalities, live births.

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I. Introduction

Congenital anomalies affect 2–3% of the births worldwide. In India, these anomalies contributed as fifth cause for neonatal deaths in 2010. (1) Neural tube defects (NTD) are the common malformations of the central nervous system, which can affect 1-2 infants per 1000 births. (2) The prevalence of NTD in some countries is estimated at 0.5-0.8/1000 births whereas; some regions have 20 times higher prevalence.(3)

Spina bifida is a birth defect and it consists of disorders (meningocele, meningomyelocele, myelocele, myelomeningocele, and rachischisis) that occur due to incomplete closure of the spinal column resulting in herniation of the spinal cord or meninges. Clinically NTD can be present as “open or cystica” type in which the affected nervous tissues are exposed or “close or occulta,” type in which the tissues are covered by skin. Mostly Open NTD form is present. (1) Spina bifida defects have lower case fatality rate than other NTD but it can lead to severe morbidity. (2) Defect with meninges protrusion is called meningocele and nervous tissue herniation results in meningomyelocele. Open NTD may occur in association with Arnold Chiari malformation, hydrocephalous and genetic syndromes, including Meckel’s syndrome, Anterior sacral meningocele, Currarino syndrome, and anal stenosis. (3) According to Sawan and Thompson, spina bifida takes place during 24 and 26 days of gestation. Spina bifida may be classified anatomically into the open or closed type and embryologically into; an error during primary or secondary neurulation. (4)

The causes of NTDs includes both environmental and genetic factors. (5) Reduced folate level is the known risk factor for NTD. (3) Non-genetic factors along with predisposing genotype effect neural tube closure. Other environmental factors include geography, epidemic trends, socio-economic level, fungal product fumonisin, antiepileptic drugs like valproic acid, hypervitaminosis A, pollutants and personal toxicants. Maternal risk factors are age, diet, diabetes and obesity. Previous studies supports genetic and chromosomal abnormalities, mostly trisomy 13 and trisomy 18 to these defects. (3,5,4) Genetic factors include rare missense mutations, variants of genes in the planar cell polarity pathway and genes encoding enzymes of folate one-carbon metabolism. (3) The mutation of the CBS gene leads to hyper-homocysteinemia have been associated to NTD. (4)

Myelomeningocele (MMC; open spina bifida) is the main form of spina bifida with brain malformations. Attention-deficit with tectal deformities are common in MMC. These neurocognitive problems can be observed at the age of 6 months. (3)

Prenatal diagnostic investigation comprised of raised alpha-fetoprotein level in amniotic fluid for anencephaly or MMC, assay of acetylcholinesterase in amniotic fluid. Biochemical screening can be advised for maternal obesity; where it impairs detailed ultrasound examination. (3) Sonographic prenatal detection of open spina bifida has higher detection rates than closed. (2) The fetal spine can be examined by ultrasonography from the late first trimester onwards. Second trimester ultrasound detects around 90-98% of MMC. (3) Recently, very few studies have been done to evaluate spina bifida in the South Indian population. Although it is commonly occurring congenital defect and recent literature shows very few study in south Indian population for NTD. Thus, our study evaluates the prevalence of spina bifida in live birth deliveries along with demographic data associated with it from a tertiary level hospital of South India. This study will provide information regarding frequency of the reported cases of the spina bifida, and further research requirements for the same.

II. Materials And Methods

The present study is a descriptive cross-sectional study, which includes retrospective analysis of demographic data for spina bifida. The study includes the number of live births deliveries in GDMCH, Tamil Nadu, India, for six years from the year 2015 to 2020. Dharmapuri district is one of the most backward districts in Tamil Nadu. (7) Ethical committee approval has been taken from the institutional ethical committee for this study. The data includes the number of live births, the number of congenital anomalies, sex of the babies, consanguineous marriage history and age of the parents, and any other sibling who also affected by same disease. Prevalence of the spina bifida cases was calculated based on the total number of live birth cases diagnosed with spina bifida at the time of birth in six years duration. Above mentioned data was entered in an MS excel sheet and statistical analysis was done based on entered data.

III. Result

In the year 2015, the total number of live births was 7263. In this, 137 cases had some congenital anomalies which are 1.8 percent of total deliveries. 11 cases had spina bifida, which is 0.2 percent of total live births and 8 percent of congenital anomalies. Out of 11 cases, 8 were female, and 3 were male. Data were obtained for 9 children, which showed that the number of cases for first child delivery was 5 and 2 cases each for second and third child delivery.

In the year 2016, the total number of live births was 8501. In this, 113 cases had some congenital anomalies which is 1.4 percent of total deliveries. 16 cases had spina bifida, which is 0.2 percent of total live births and 14 percent of total congenital anomalies. Out of 16 cases, 7 were female and 9 were male. Data for 8 children showed that the number of cases for first child delivery was 5, second child delivery was 2, and third child delivery was 1.

In the year 2017, the total number of live births was 7898. In this, 103 cases had some congenital anomalies, which is 1.4 percent of total deliveries. 7 cases had spina bifida, which is 0.1 percent of total live births and 7 percent of total congenital anomalies. Out of 7 cases, 2 were female and 5 were male. The number of cases for first child delivery was 3; second child delivery was 2, and 2 for third child delivery.

In the year 2018, the total number of live births was 7990. In this, 122 cases had some congenital anomalies, which is 1.9 percent of total deliveries. 8 cases had spina bifida, which is 0.1 percent of total live births and 7 percent of total congenital anomalies. Out of 8 cases, 3 were female and 5 were male. The number of cases for first child delivery was 3, second child delivery was 2, and 3 for third child delivery.

In the year 2019, the total number of live births was 9473. In this 206 cases had some congenital anomalies, which is 2.2 percent of total deliveries. 8 cases had spina bifida, which is 0.1 percent of total live births and 7 percent of total congenital anomalies. Out of 8 cases, 5 were female and 3 were male. Number of cases for first child delivery was 3, and second child delivery was 5.

In the year 2020, the total number of live births was 10,239. In this, 384 cases had some congenital anomalies, which is 3.7 percent of total deliveries. 16 cases had spina bifida, which is 0.1 percent of total live births and 4 percent of total congenital anomalies. Out of 16 cases, 8 were female, 8 were male and 1 transgender. The number of cases for first child delivery was 10, second child delivery was 4 and third child delivery was 2.

Prevalence of Spina Bifida cases in live births was calculated by the following formula (8)

Number of spina bifida cases in live births reported in 6 years duration x 1000
Total number of live birth deliveries conducted during a time period

$$=66 \times 1000 / 51364 = 1.28$$

IV. Discussion:

51364 live births occurred during the years 2015 to 2020. Total number of congenital defects are 1065 and 66 cases of spina bifida were identified clinically immediately after birth that is 6.19 % out of total defects in six years duration. (Figure-1) The live birth prevalence of spina bifida was 1.28 per 1000. Telephonically we were able to contact 56 cases out of 66 cases.

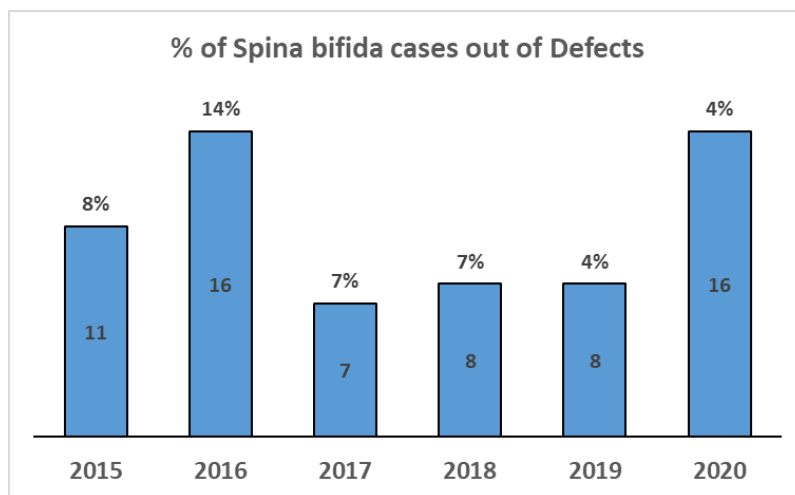


Figure 1: Percentage of spina bifida out of total congenital defects.

Meningocele case by sex: Total number of spina bifida cases reported in live births were 66, out of that 33 patients were female from 24472 live births and 33 patients were male from 26891 live births. Prevalence for girls was 1.34 /1000 and Prevalence for males was 1.227 /1000. From 66 cases, 6 males and 7 females could not survive post delivery. Thus, around 19.70 percent of the patients expired with a history of spina bifida. Thus, the ratio for the number of cases for males and females was equal. (Figure2,3)

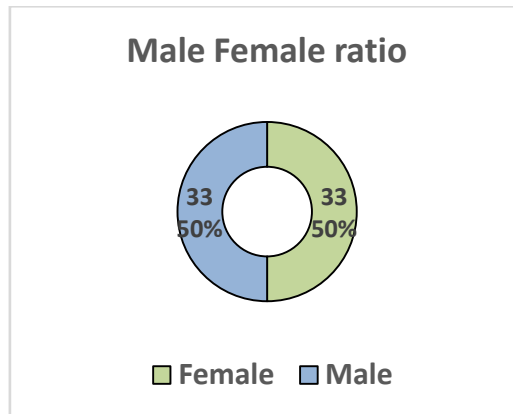


Figure2: Male female ratio for spina bifida

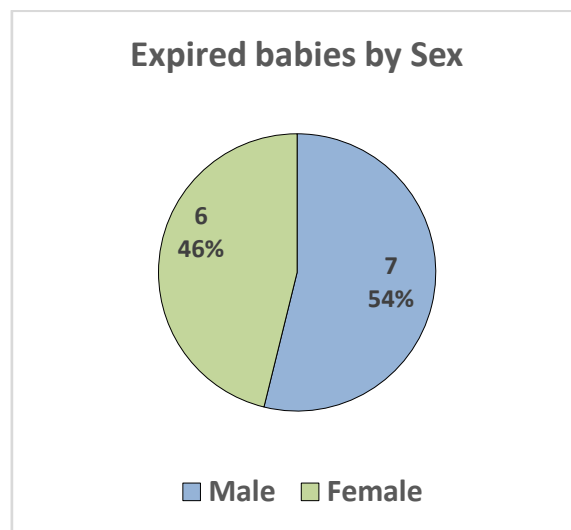


Figure3: Number of expired babies after delivery.

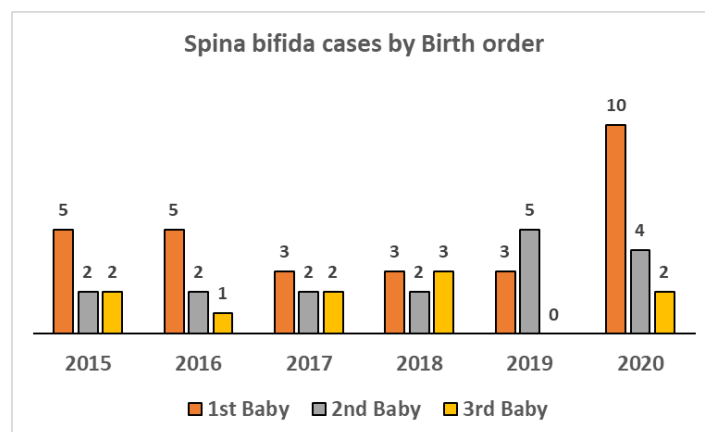


Figure4: Spina bifida cases by birth order.

Spina bifida cases by birth order- 29 cases out of 56 cases were of first baby pregnancy, 19 cases were of second baby delivery and 10 cases for third baby delivery . Thus, almost 51.7 % 33.9% and 17.8% were from first, second and third baby delivery respectively . (Figure-4) Parent's age for spina bifida cases-Age of the parent's was divided into following 4 groups based on data . Father's age group: 18-20 years- Nil, 21-30 years- 28, 31-40 years-27 and 1-50 years-1. Most commonly Father's age were in the age group of 21-40 years .

Mother's age group: 18-20 years- 9, 21-30 years- 44, 31-40 years-03 and 1-50 years-Nil. Most of the mothers were in the age range of 21-30 years. Consanguineous marriage history: 56 patients were contacted telephonically and 24 parents gave positive history of consanguineous marriage and 32 parents had negative history .

Globally an incidence of 1.0/1000 live births for NTD's have been reported by Lemire. (8) A systematic review done in India in 2013 showed 1310 cases of the neural tube defects in 308,387 births, giving an overall birth prevalence of 4.1 per 1000. The live birth and stillbirth prevalence of neural tube defects were 1.3 per 1000 births and 1.7 per 1000 births, respectively. Among the neural tube defects, prevalence for spina bifida is 1.9 per 1000 births. (9) A population based survey in North Indian population showed total of 26,946 live births occurred during the years 2001 to 2014 and a total of 140 confirmed cases of NTDs were identified. The live birth prevalence of NTDs was 24.1 per 10,000 live births. Spina bifida was the most common type of NTD followed by anencephaly and encephalocele. (10) While our study showed prevalence of 1.28 per 1000 spina bifida live births in GDMCH. Same percentage of cases were observed for male and female patients and almost 19.7% of the cases could not survive beyond six years. 42.8% of the patient's parents gave history of consanguineous marriage. 51.7% of the cases were for first baby delivery. Almost 6% of the cases out of the total reported congenital anomalies were spina bifida.

Preconceptional administration of 400µg folic acid results in the reduction of 60% to 70% NTDs occurrence. (3) Mandatory folic acid fortification showed lower period prevalence estimates of spina bifida in live births, stillbirths and termination of the pregnancy. (2) Pregnant women having 'High risk' for NTD can take 4mg folic acid, whereas those at low risk are advised to take 0.4 mg. The underlying mechanisms which reduce neural tube defects risk remain unexplained. Inositol has shown encouraging results but needs further research. Supplements containing vitamin B12, a co-factor in folate one-carbon metabolism, may further reduce the frequency of NTDs. Some may not respond to exogenous folic acid owing to defects in the metabolic enzymes. In this case, alternative folates, such as 5-methyl tetrahydrofolate can be considered.

The management of MMC involves surgery within 48 hours of birth. The child's back is closed to minimize the risk of infection that can result in meningitis. Orthopedic deformities are usually treated after birth. Management of spina bifida is best provided by a multidisciplinary team. Early repair of the lesion, in utero, may prevent continuing damage. There is risk of premature birth in such cases. About 25% of mothers in the fetal surgery demonstrated thinning of the uterine wall during cesarean delivery, and 10% showed partial (9%) or complete (1%) tissue edge separation at the hysterotomy site. A mother carrying a fetus with MMC, at less than 24 weeks gestation has three choices: termination of pregnancy, continuation of the pregnancy with near-term cesarean section and postnatal repair, or prenatal surgery. (3)

V. Conclusion

Conclusion: Spina bifida is one of the commonly occurring congenital neural tube defects, which could be associated with severe comorbidities. Reduced incidence of these defects occur with preconceptual fortification of folic acid. Spina bifida can be diagnosed at an early stage by conducting ultrasound screening during early pregnancy. National registry of the congenital neural tube anomalies at the time of birth is advised for understanding the prevalence of various birth defects and need for further research. The data will specify the impact of congenital anomaly affected pregnancies or births on health service employment, for deciding either termination of pregnancy or for required neonatal intensive care services. Some of these conditions can be prevented through primary care involvement directed towards women in the preconception, intra-conception and antenatal periods like mandatory preconceptual folic acid fortification and awareness about maternal care during pregnancy.

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